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A Case of Metabolic Genetic Emergency

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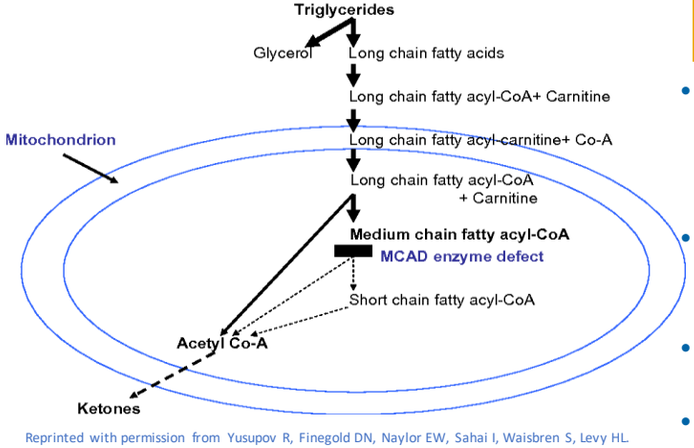
The Case

- 4-day old infant presents to pediatrician for follow up
- Prenatal history unremarkable, born to a 34-year-old G3P2 via C section for non-reassuring fetal heart tones, briefly required CPAP at delivery, normal nursery course, breastfeeding on demand
- Found to exhibit lethargy, hypoglycemia, hypothermia, sunken fontanelle, 8% below birthweight
- CBCd, BMP, LFTs unremarkable aside from glucose 45, slight elevation in ALT to 72
- Feeding observation: poor swallow-suck-breathe coordination, difficulty pacing
- Differential diagnosis includes: sepsis: bacterial vs viral, HSV; inborn error of metabolism; congenital heart disease; hypothyroidism; inadequate intake

The pathway of mitochondrial fatty acid ß-oxidation



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Diagnosis and Discussion

- On the same day of presentation, newborn screen confirmed diagnosis of medium chain acyl CoA dehydrogenase deficiency (MCADD)
- Disorder of fatty acid beta oxidation, cannot convert medium chain fatty acyl CoA into short chain and ketones in times of fasting
- Affects 1 in 9,000-20,000 newborns, autosomal recessive mutation in ACADM gene
- Confirm by acylcarnitine profile showing elevation of C8 and urine organic acids
- Prognosis excellent when detected on newborn screen prior to symptoms
- Treatment: reducing fasting intervals (no longer than 2-3 hours until 4mo)
- Emergency plan: fluids with 10g of sugar per 100mL, or admission with D10 at 1.5x maintenance
- More info on inborn errors: ACTion sheets (ACMG)

Conclusions

- Inborn errors of metabolism must be high on the differential in a neonate with hypoglycemia and/or signs and symptoms of sepsis in conjunction with poor feeding
- MCAD deficiency is a devastating disease screened for universally on day of life 1, and must be acted upon promptly when identified on newborn screen
- Confirmation of diagnosis includes acylcarnitine profile and genetic evaluation
- Treatment includes avoidance of fasting, emergency plan for illness

References

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